Red Flags of Familial Hypercholesterolemia

The following are family and personal history signs that may suggest familial hypercholesterolemia (FH). They are based on guidelines from the National Institute for Health and Care Excellence (NICE) in the U.K. and the Simon Broome Diagnostic Criteria, as well as guidelines from the U.S. National Lipid Association.

Y/N	Red Flag	Additional Information
	Early age of onset for coronary artery disease in self or in family (includes heart attack, stent, bypass)	For men< 50 years old For women < 60 years old (Simon Broome Criteria) For men < 55 years old For women < 65 years old (National Lipid Association)
	Family history of sudden death	For men< 50 years For women < 60 years old (Simon Broome Criteria) For men < 55 years For women < 65 years old (National Lipid Association)
	Total cholesterol ≥220 mg/dL or LDL cholesterol ≥190 mg/dL in individual	Should be untreated and fasting cholesterol levels; other causes of hyperlipidemia should be ruled out (NICE)
	Tendon xanthomas	Typically found on the Achilles tendon, and finger extensor (NICE, National Lipid Association)

If your patient has even one "Yes", they are **suspected** of having FH.

Next Steps:

- At least a three-generation pedigree that includes the following:
  - Age of onset of coronary heart disease
  - Lipid concentrations
  - Smoking history
  - If deceased, cause and age of death
- Cholesterol screening (if it was not done previously)
  - Patient should be fasting and not on medication
  - Rule out other causes of hypercholesterolemia (i.e. diet, lack of exercise)
- Referral to a cardiovascular genetics specialist for consideration of genetic testing

For additional information, please see:

- http://guidance.nice.org.uk/cg71
- http://www.ncbi.nlm.nih.gov/books/NBK53810/
- Goldberg AC, Hopkins PN, Toth PP, et al. Familial Hypercholesterolemia: Screening, diagnosis and management of pediatric and adult patients. J Clin Lipidol. 2011;5:133-140.

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